MOVEMENT DISORDERS GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype Description and OMIM disease ID
AARS2	132.9	99%	98%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCB7	156	99%	97%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	96	77%	68%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	114.9	97%	91%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	134.1	98%	93%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADAR	131.7	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADCK3	146	99%	98%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCY5	144.8	93%	91%	Dyskinesia, familial, with facial myokymia, 606703
AFG3L2	126.4	92%	85%	Ataxia, spastic, 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
ALDH18A1	143	100%	99%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH3A2	157.4	100%	99%	Sjogren-Larsson syndrome, 270200
ALS2	184.7	99%	99%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ANO10	126.6	99%	96%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	175	99%	97%	Dystonia 24, 615034
AP4B1	166	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	117.3	99%	96%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	125.9	99%	97%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78.3	74%	68%	Spastic paraplegia 52, autosomal recessive, 614067

AP5Z1	95	99%	98%	Spastic paraplegia 48, autosomal recessive, 613647
APTX	136.6	93%	90%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	172.1	100%	100%	Argininemia, 207800
ARSA	111.5	100%	99%	Metachromatic leukodystrophy, 250100
ARX	39.1	82%	70%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASPA	151.7	99%	92%	Canavan disease, 271900
ATCAY	166.6	100%	99%	Ataxia, cerebellar, Cayman type, 601238
ATL1	195.9	98%	95%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATM	124.3	98%	93%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
ATP13A2	129	99%	98%	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP1A2	209.6	100%	100%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	205.7	100%	100%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2B3	161.5	99%	98%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	173	99%	99%	Wilson disease, 277900
B4GALNT1	163.9	99%	96%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	74.1	93%	81%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	186.9	99%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	124.4	89%	81%	Maple syrup urine disease, type Ib, 248600
BSCL2	126.4	100%	99%	Encephalopathy, progressive, with or without lipodystrophy, 615924

				Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
C10orf2	193.6	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
C12orf65	91.3	97%	92%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	100.8	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CA8	120.5	95%	90%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	105.6	95%	91%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1G	147.8	99%	98%	Spinocerebellar ataxia 42, 616795
CACNB4	126.8	98%	97%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CAMTA1	193.2	99%	99%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN1	169.4	100%	100%	Spastic paraplegia 76, autosomal recessive, 616907
CCT5	166	100%	99%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	176.8	99%	97%	No OMIM phenotype Cervical dystonia, primary (Xiao (2012) Ann Neurol 71, 458)
COASY	168	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COL4A1	101.8	98%	93%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COQ2	84.5	95%	92%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ9	105.7	99%	98%	Coenzyme Q10 deficiency, primary, 5, 614654
COX20	52.3	90%	73%	Mitochondrial complex IV deficiency, 220110

CP	141	94%	90%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CSF1R	144.4	99%	98%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSTB	120.6	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	188.7	97%	95%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	140.9	95%	92%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	106.9	95%	90%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	122.5	97%	92%	Maple syrup urine disease, type II, 248600
DCAF17	110.1	98%	92%	Woodhouse-Sakati syndrome, 241080
DCTN1	143.1	99%	99%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DDC	118.6	99%	96%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	166.1	96%	94%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	184.8	99%	96%	Spastic paraplegia 54, autosomal recessive, 615033
DLAT	102.4	99%	95%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	142	99%	97%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNMT1	129	99%	98%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
EIF2B1	156.3	100%	99%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	157.2	100%	99%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	180	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	161.8	99%	99%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	133.8	99%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	144.4	99%	99%	{Parkinson disease 18}, 614251
ELOVL5	129.5	100%	99%	Spinocerebellar ataxia 38, 615957
ERLIN2	167.7	99%	99%	Spastic paraplegia 18, autosomal recessive, 611225
FA2H	108.9	94%	88%	Spastic paraplegia 35, autosomal recessive, 612319
FAR1	91.8	96%	91%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154

FBXO7	237.5	99%	97%	Parkinson disease 15, autosomal recessive, 260300
FGF14	219	99%	98%	Spinocerebellar ataxia 27, 609307
FLVCR1	149.3	99%	96%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOLR1	166.3	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD7	144.2	99%	98%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	131.3	99%	92%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GALC	112.3	97%	93%	Krabbe disease, 245200
GAN	210.8	99%	99%	Giant axonal neuropathy-1, 256850
GBA	237.3	100%	100%	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	187.3	100%	99%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	148.6	93%	91%	Glutaricaciduria, type I, 231670
GCH1	91.2	95%	86%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	110.3	99%	98%	Alexander disease, 203450
GJC2	52.2	85%	68%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLB1	93.9	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	153.7	95%	92%	Dystonia 25, 615073
GOSR2	143.1	97%	95%	Epilepsy, progressive myoclonic 6, 614018
GPR143	77.7	91%	85%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500

GPR56	173.8	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
GRID2	195.3	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRM1	194.5	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
HEXB	152.6	97%	91%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	75.3	94%	84%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSPD1	92.7	96%	89%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
ITPR1	176.7	100%	99%	Gillespie syndrome,206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	178.7	100%	99%	Episodic ataxia/myokymia syndrome, 160120
KCNC1	213.9	100%	99%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	155.3	71%	58%	Spinocerebellar ataxia 13, 605259
KCND3	207.2	100%	98%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNJ10	229	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ6	196.9	100%	99%	Keppen-Lubinsky syndrome, 614098
KCNMA1	158.7	100%	99%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCTD7	144.9	93%	92%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA0196	159.1	98%	96%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
KIAA0226	116.4	98%	97%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA2022	183	100%	99%	Mental retardation, X-linked 98, 300912
KIF1A	134.8	99%	97%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	126.4	99%	99%	Spastic ataxia 2, autosomal recessive, 611302
KIF5A	147.5	100%	99%	Spastic paraplegia 10, autosomal dominant, 604187
KMT2B	135.2	94%	91%	No OMIM phenotype Kleefstra-like syndrome (Agha (2014) PLoS One 9,e112687)
L1CAM	157.7	99%	98%	Corpus callosum, partial agenesis of, 304100

				CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
MARS2	168.8	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MECP2	100	99%	94%	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MICU1	140	95%	91%	Myopathy with extrapyramidal signs, 615673
MMADHC	82.8	87%	74%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MRE11A	57.6	95%	85%	Ataxia-telangiectasia-like disorder, 604391
MTHFR	153.2	100%	99%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTPAP	133	98%	93%	Ataxia, spastic, 4, 613672
MTTP	155.8	99%	98%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
NIPA1	173.4	99%	99%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	55.6	98%	93%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550
NOL3	95	94%	87%	Myoclonus, familial cortical, 614937
NPC1	162.1	99%	98%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}

NPC2	144	100%	99%	Niemann-pick disease, type C2, 607625
NUP62	126.9	99%	99%	Striatonigral degeneration, infantile, 271930
OPA1	135.3	98%	91%	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
PANK2	177.5	99%	96%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX6	156.1	100%	99%	Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229 ?Morning glory disc anomaly, 120430
PDE10A	160.6	99%	99%	Dyskinesia,limb and orofacial,infantile-onset,616921 Striatal degeneration,autosomal dominant,616922
PDE8B	121.8	99%	98%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	107.8	100%	99%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	166.4	99%	97%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDHA1	127.8	97%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	136.1	98%	96%	Lacticacidemia due to PDX1 deficiency,245349
PDSS1	134.8	91%	85%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	131.3	97%	93%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	121.7	100%	100%	Spinocerebellar ataxia 23, 610245

PEX10	118.3	97%	93%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX7	138.5	89%	85%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	86.5	98%	92%	Refsum disease, 266500
PIK3R5	123.1	100%	99%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	132.4	99%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLP1	162.1	100%	99%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	178.4	99%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNKD	107.7	99%	98%	Paroxysmal nonkinesigenic dyskinesia,118800
PNKP	98.4	99%	97%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPLA6	140.9	99%	98%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
POLG	126.2	99%	99%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR3A	162.2	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	168.9	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
PRKCG	127.5	97%	93%	Spinocerebellar ataxia 14, 605361
PRKRA	161.3	99%	99%	Dystonia 16, 612067
PRRT2	80.7	99%	98%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PYCR2	137.6	99%	98%	Leukodystrophy, hypomyelinating, 10, 616420

REEP1	113.8	98%	96%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
RNASEH2A	149.3	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	125.1	94%	84%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	207.6	99%	97%	Aicardi-Goutieres syndrome 3, 610329
RNF170	149.4	98%	93%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF216	154.3	99%	98%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RTN2	113.2	98%	95%	Spastic paraplegia 12, autosomal dominant, 604805
SACS	170.8	100%	99%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	149.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SCN8A	224.3	99%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SERAC1	125.5	98%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	187.1	99%	99%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SGCE	95.4	94%	89%	Dystonia-11, myoclonic, 159900
SIL1	173.8	99%	98%	Marinesco-Sjogren syndrome, 248800
SLC12A6	169.6	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	69.6	96%	86%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	191.3	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	153.6	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	122.2	99%	95%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	228.7	98%	95%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC2A1	183.5	100%	100%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC30A10	192.1	99%	99%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	148.5	96%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC39A14	122.3	99%	98%	Hypermanganesemia with dystonia 2, 617013

SLC52A2	196.4	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	124.5	100%	99%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC6A3	153.5	100%	99%	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC9A1	169.6	100%	100%	?Lichtenstein-Knorr syndrome, 616291
SMPD1	134.3	99%	97%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNCA	150	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNX14	76.2	92%	82%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SPAST	71.8	93%	82%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	146.9	98%	96%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG20	166.4	99%	97%	Troyer syndrome, 275900
SPG21	146.1	99%	96%	Mast syndrome, 248900
SPG7	127.9	96%	92%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	183.9	97%	89%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	122.1	99%	99%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	174	99%	99%	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUOX	219.5	100%	100%	Sulfite oxidase deficiency, 272300
SYNE1	156.8	99%	99%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
TAF1	141.1	99%	97%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TDP1	132	98%	95%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TECPR2	165.7	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TGM6	153.7	99%	97%	Spinocerebellar ataxia 35, 613908
TH	83.6	97%	92%	Segawa syndrome, recessive, 605407
THAP1	152.3	100%	100%	Dystonia 6, torsion, 602629
TIMM8A	45.5	87%	70%	Jensen syndrome, 311150

				Mohr-Tranebjaerg syndrome, 304700
TMEM240	130.5	100%	99%	Spinocerebellar ataxia 21, 607454
TMEM67	78.9	92%	83%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOR1A	233.5	100%	99%	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}
TPP1	158.7	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREM2	134.3	99%	98%	Nasu-Hakola disease, 221770
TREX1	272.2	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TTBK2	169.4	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	106.2	90%	81%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	122.7	93%	84%	Ataxia with isolated vitamin E deficiency, 277460
TUBB4A	137	96%	95%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	185.9	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TYROBP	95.1	100%	100%	Nasu-Hakola disease, 221770
VAMP1	156.3	100%	100%	Spastic ataxia 1, autosomal dominant, 108600
VCP	168.4	99%	99%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VLDLR	244.5	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	81	93%	84%	Choreoacanthocytosis, 200150
VPS37A	79.7	86%	69%	Spastic paraplegia 53, autosomal recessive, 614898
VRK1	147	98%	95%	Pontocerebellar hypoplasia type 1A, 607596
WDR45	92.9	97%	92%	Neurodegeneration with brain iron acculation 5, 300894
WDR81	171.3	99%	99%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	148	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211

				Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XPR1	155.9	99%	99%	Basal ganglia calcification, idiopathic, 6, 616413
ZFYVE26	134.1	99%	99%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	137.1	100%	100%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	140.6	100%	99%	No OMIM phenotype

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors